



## Bietti crystalline dystrophy

Bietti crystalline dystrophy is a disorder in which numerous small, yellow or white crystal-like deposits of fatty (lipid) compounds accumulate in the light-sensitive tissue that lines the back of the eye (the retina). The deposits damage the retina, resulting in progressive vision loss.

People with Bietti crystalline dystrophy typically begin noticing vision problems in their teens or twenties. They experience a loss of sharp vision (reduction in visual acuity) and difficulty seeing in dim light (night blindness). They usually lose areas of vision (visual field loss), most often side (peripheral) vision. Color vision may also be impaired.

The vision problems may worsen at different rates in each eye, and the severity and progression of symptoms varies widely among affected individuals, even within the same family. However, most people with this condition become legally blind by their forties or fifties. Most affected individuals retain some degree of vision, usually in the center of the visual field, although it is typically blurry and cannot be corrected by glasses or contact lenses. Vision impairment that cannot be improved with corrective lenses is called low vision.

### Frequency

Bietti crystalline dystrophy has been estimated to occur in 1 in 67,000 people. It is more common in people of East Asian descent, especially those of Chinese and Japanese background. Researchers suggest that Bietti crystalline dystrophy may be underdiagnosed because its symptoms are similar to those of other eye disorders that progressively damage the retina.

### Genetic Changes

Bietti crystalline dystrophy is caused by mutations in the *CYP4V2* gene. This gene provides instructions for making a member of the cytochrome P450 family of enzymes. These enzymes are involved in the formation and breakdown of various molecules and chemicals within cells. The *CYP4V2* enzyme is involved in a multi-step process called fatty acid oxidation in which lipids are broken down and converted into energy, but the enzyme's specific function is not well understood. *CYP4V2* gene mutations that cause Bietti crystalline dystrophy impair or eliminate the function of this enzyme and are believed to affect lipid breakdown. However, it is unknown how they lead to the specific signs and symptoms of Bietti crystalline dystrophy. For unknown reasons, the severity of the signs and symptoms differs significantly among individuals with the same *CYP4V2* gene mutation.

## **Inheritance Pattern**

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- BCD
- Bietti crystalline corneoretinal dystrophy
- Bietti crystalline retinopathy
- Bietti tapetoretinal degeneration with marginal corneal dystrophy

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Bietti crystalline corneoretinal dystrophy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1859486/>

### Other Diagnosis and Management Resources

- GeneReview: Bietti Crystalline Dystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK91457>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Retinal Disorders  
<https://medlineplus.gov/retinaldisorders.html>

### Genetic and Rare Diseases Information Center

- Bietti crystalline corneoretinal dystrophy  
<https://rarediseases.info.nih.gov/diseases/10050/bietti-crystalline-corneoretinal-dystrophy>

### Additional NIH Resources

- National Eye Institute: Facts About Bietti's Crystalline Dystrophy  
<https://nei.nih.gov/health/biettis/bietti>
- National Eye Institute: Low Vision  
<https://nei.nih.gov/lowvision/>

### Educational Resources

- American Academy of Ophthalmology: What is Low Vision?  
<https://www.aao.org/eye-health/diseases/low-vision>
- American Optometric Association: Low Vision  
<http://www.aoa.org/patients-and-public/caring-for-your-vision/low-vision?sso=y>
- Disease InfoSearch: Bietti Crystalline Corneoretinal Dystrophy  
<http://www.diseaseinfosearch.org/Bietti+Crystalline+Corneoretinal+Dystrophy/825>
- MalaCards: bietti crystalline corneoretinal dystrophy  
[http://www.malacards.org/card/bietti\\_crystalline\\_corneoretinal\\_dystrophy](http://www.malacards.org/card/bietti_crystalline_corneoretinal_dystrophy)
- Orphanet: Bietti crystalline dystrophy  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=41751](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=41751)
- The University of Arizona College of Medicine  
<http://disorders.eyes.arizona.edu/handouts/bietti-crystalline-corneoretinal-dystrophy>

### Patient Support and Advocacy Resources

- American Foundation for the Blind: Low Vision  
<http://www.visionaware.org/info/your-eye-condition/eye-health/low-vision/123>
- Retina International  
<http://www.retina-international.org/>

### GeneReviews

- Bietti Crystalline Dystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK91457>

### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22Bietti+crystalline+dystrophy%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28bietti+crystalline+dystrophy%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- BIETTI CRYSTALLINE CORNEORETINAL DYSTROPHY  
<http://omim.org/entry/210370>

## **Sources for This Summary**

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